
Allele-specific methylation is prevalent and is contributed by CpG-SNPs in the human genome.

Journal: Genome Res

Publication Year: 2010

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PubMed link: 20418490

Funding Grants: Interdisciplinary Stem Cell Training Program at UCSD II

Public Summary:

Scientific Abstract:

In diploid mammalian genomes, parental alleles can exhibit different methylation patterns (allele-specific DNA methylation, ASM), which have been documented in a small number of cases except for the imprinted regions and X chromosomes in females. We carried out a chromosome-wide survey of ASM across 16 human pluripotent and adult cell lines using Illumina bisulfite sequencing. We applied the principle of linkage disequilibrium (LD) analysis to characterize the correlation of methylation between adjacent CpG sites on single DNA molecules, and also investigated the correlation between CpG methylation and single nucleotide polymorphisms (SNPs). We observed ASM on 23% approximately 37% heterozygous SNPs in any given cell line. ASM is often cell-type-specific. Furthermore, we found that a significant fraction (38%-88%) of ASM regions is dependent on the presence of heterozygous SNPs in CpG dinucleotides that disrupt their methylation potential. This study identified distinct types of ASM across many cell types and suggests a potential role for CpG-SNP in connecting genetic variation with the epigenome.

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